Deletion 22q11.2 Syndrome

What is it?

Deletion 22q11.2 Syndrome is a congenital genetic disorder that occurs when a small part of chromosome 22 is missing or deleted (22q11.2). **Chromosomes** are located in the cells in our body. Typically, people are born with 23 pairs of chromosomes. **Genes** (the body's blueprint) are located inside the chromosomes and contain our **DNA** (carrier of our genetic information). The 22q11.2 deletion can cause multiple health issues, including DiGeorge syndrome, velocardiofacial syndrome, conotruncal anatomy face syndrome, Opitz G/BBB, and Cayler cardiofacial syndromes. It is a common genetic disorder that occurs in about 1 out of every 4,000 births each year. About 5%-8% of these infants have a cleft palate. Some infants are diagnosed at birth, but some may not display symptoms until 1-3 years of age or older. Symptoms can range from mild to severe.

Common symptoms include:

- heart problems
- low muscular tone
- speech difficulties
- middle ear infections or hearing loss
- vision problems
- feeding problems
- frequent infections
- learning disorders, especially with visual materials
- developmental delays
- communication and social interaction problems
- psychiatric issues

Facial features may include:

- small ears with squared upper ear
- hooded eyelids
- cleft palate
- asymmetric facial appearance when crying
- small mouth, chin, and side areas of the nose tip

Table 1

How common is it?

It is a common genetic disorder that occurs in about 1 out of every 4,000 births each year.

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What causes it?

Deletion 22q11.2 syndrome occurs when a small part of chromosome 22 is deleted or missing (22q11.2). This deletion is usually a random occurrence and can take place during the development of the fetus. It is rare for this condition to be inherited. About 5%-10% of infants inherit the disorder from a parent who has a mild case.

How is it diagnosed?

The deletion 22q11.2 syndrome diagnosis is confirmed by a blood test called a chromosomal analysis. **Chromosomal analysis** is a test that looks at and determines the number of chromosomes in a person.

How is it treated?

There is no cure for deletion 22q11.2 syndrome. Our genetics cannot be altered. However, there are treatment options available for different health issues associated with the syndrome. These will differ for each person depending upon their symptoms. Your child's doctor will discuss appropriate treatment options with you.

For more information:

Boston Children's Hospital
https://www.childrenshospital.org/conditions-and-treatments/conditions/d/22q11-2-deletion-syndrome/diagnosis-and-treatment

Children's Hospital of Philadelphia (Table 1)
https://www.chop.edu/conditions-diseases/22q112-deletion-and-duplication-syndromes

Mayo Clinic
https://www.mayoclinic.org/diseases-conditions/digeorge-syndrome/diagnosis-treatment/drc-20353548

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